CLAIMS

That which is claimed is:

1. A method of prenatal diagnosis of a fetus affected by Smith-Lemli-Optiz syndrome (SLOS) comprising:

obtaining a biological sample from a woman suspected of carrying an SLOS-affected fetus;

analyzing the sample for a at least one specific SLOS analyte and a control steroid counterpart, wherein the SLOS analyte is selected from the group consisting of 8-dehydro-estriol (8-DHE3) and 5β-pregn-7-ene-α,17 α,20 α-triol (7-DHPT); and

determining the ratio of the SLOS analyte to the control steroid counterpart found in the sample;

wherein a ratio of greater than 0.01 indicates that the woman carries an SLOS-affected fetus.

2. The method of claim 1, wherein the biological sample is a urine sample.

3. The method of claim 1, wherein the biological sample is serum.

4. The method of claim 1, wherein the sample is a blood or blood-derived sample.

5. The method of claim 1, wherein said analyzing comprises analysis by use of gas chromatography-mass spectrometry.

6. The method of claim 1, wherein said analyzing is by use of high-performance liquid chromatography/mass spectrometry.

7. The method of claim 1, wherein the biological sample is obtained as early as about the 11th week of gestation.

8. The method of claim 1, wherein the SLOS analyte is 8-DHE3 and the steroid counterpart is a estriol (E_3).

9. The method of claim 1, wherein the SLOS analyte is a 7-DHPT and the control steroid counterpart is a 5β-pregnane-3α,17 α,20 α-triol (PT).

10. The method of claim 1, wherein each of the SLOS analytes 8-DHE_3 and 7-DHPT are detected.